

# **PARENT FACT SHEET**

## **DISORDER**

### **Propionic Acidemia (PA)**

#### **CAUSE**

Propionic Acidemia (PA) is one type of organic acid disorder. People with PA have problems breaking down and using certain amino acids from the foods they eat. PA occurs when an enzyme called “propionyl CoA carboxylase” is either missing or not working properly. When this enzyme is not working, substances called glycine and Propionic acid along with other harmful substances build up in the blood and cause problems.

#### **IF NOT TREATED**

The symptoms can start as early as the first week of life. If untreated, harmful substances from the body spill into the blood and urine. This is called a metabolic acidosis. If a metabolic crisis is not treated, a child with PA can develop breathing problems, stroke, swelling of the brain, seizures and coma-sometimes leading to death.

A metabolic crisis can be triggered by eating large amounts of protein, illness or infection, going to long without food and stressful events such as surgery.

#### **TREATMENT OPTIONS**

- Your doctor will work with a metabolic specialist and dietitian to care for your child. Lifelong treatment is usually needed.
- Avoid going a long time without food. These babies need to eat more often to avoid low blood sugar. They should not go without eating for more than 4-6 hours. Some babies will need to eat more often than this.
- A low protein diet is often recommended. Your dietitian will help plan any diet changes.
- Medical formula and foods contain the correct amount of protein and nutrients needed to for normal growth and development. Your metabolic doctor will tell you what type of formula is best and how much to use.
- A medication that has been found to be beneficial is L-Carnitine. It is a safe and natural substance that helps the body create energy and rid the body of harmful wastes. Unless you are advised otherwise, use only L-Carnitine prescribed by your doctor. Some children may be prescribed Biotin supplements, which is a type of B vitamin.
- Contact your child’s doctor immediately at the start of any illness.

#### **IF TREATED**

It is not known how effective treatment is in preventing problems. Children who need treatment are treated early may be able to live healthy lives with typical growth and development. Some children may continue to have seizures, mental delays, involuntary movements and other health problems despite treatment.